

DEPARTMENT OF HEALTH
Rules Amending Title 11
Hawaii Administrative Rules
Date

1. Chapter 11-143, Hawaii Administrative Rules, entitled, "Testing of Newborn Infants for Metabolic and Other Diseases" is amended and compiled to read as follows:

"HAWAII ADMINISTRATIVE RULES

TITLE 11

DEPARTMENT OF HEALTH

CHAPTER 143

TESTING OF NEWBORN INFANTS FOR
METABOLIC AND OTHER DISEASES

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Historical Note: This chapter is based substantially on chapter 11-142. [Eff 09/18/81; R 1/24/87]

§11-143-1 Purpose. The purpose of this chapter is to establish requirements to assure the testing for and detection of specified metabolic and other diseases

in all infants born in the State in order to permit the institution of effective treatment for affected infants. [Eff 1/24/87; comp] (Auth: HRS §321-291) (Imp: HRS §321-291)

§11-143-2 Applicability. This chapter applies to all infants born in the State of Hawaii, to all physicians caring for newborn infants, to all persons assisting the birth of a child not attended by a physician, to all licensed laboratories and perinatal health care facilities in the State, and to the designated laboratory performing newborn screening tests. [Eff 1/24/87; comp] (Auth: HRS §321-291) (Imp: HRS §321-291)

§11-143-3 Definitions. As used in this chapter: "Acceptable specimen" means a specimen on which an accurate laboratory analysis can be performed for the disease for which it was submitted.

"Amino acid disorders" means a group of hereditary disorders caused by enzymatic defects, which result in a toxic accumulation of certain amino acids in the blood.

"Biotinidase deficiency" means a recessively inherited disease which affects the regeneration of the vitamin-cofactor biotin and impairs the metabolism of mitochondrial carboxylases.

"Birth attendant" means any person licensed or certified by the State to provide maternity care and to deliver pregnant women, or any person assisting the birth of an infant not attended by a licensed or certified practitioner. Transport personnel are excluded.

"Confirmatory specimen" means a specimen collected for the purpose of performing a confirmatory test.

"Confirmatory test" means a test performed on a specimen to determine the validity of a previous positive test.

"Congenital adrenal hyperplasia" means a group of hereditary diseases caused by an inborn error of cortisol production.

"Congenital hypothyroidism" means a disease of the thyroid gland which results in deficiency of thyroid hormone in the neonate.

"Department" means the department of health, State of Hawaii.

"Designated laboratory" means a laboratory selected by the department to perform newborn screening tests.

"Director" means the director of the state department of health.

"Fatty acid oxidation disorders" means a group of hereditary disorders caused by defects in enzymes which are involved in the breakdown of dietary and stored fats to energy.

"Galactosemia" means a disease, usually due to a single enzyme deficiency of genetic origin, which results in an abnormal increase in the concentration of galactose in the blood.

"Hemoglobinopathies" means conditions in which a mutation in the hemoglobin gene, or in genes involved in hemoglobin synthesis, produces variations in the hemoglobin structure, function, or quantity.

"Hospital" means any health facility licensed by the State and approved to provide perinatal and pediatric services.

"Initial specimen" means the first specimen collected for newborn screening.

"Kit" means materials provided by the designated laboratory for the purposes of newborn screening specimen collection and submission of specimens for newborn screening laboratory tests.

"Maple syrup urine disease" means a recessively inherited disease which is characterized by an inability to metabolize the branched chain amino acids, leucine, isoleucine, and valine.

"Negative" means a laboratory result on an acceptable specimen which is designated as having insufficient risk for disease to justify follow-up action.

"Newborn screening specimen" means a fluid or tissue collected from the newborn to be submitted for newborn screening tests.

"Newborn screening test" means a laboratory procedure performed on newborns to detect those at sufficiently increased risk for the diseases specified in section 11-143-4 to justify follow-up action.

"Organic acid disorders" means a group of hereditary disorders caused by enzymatic defects which result in a toxic accumulation of certain organic acids in the blood.

"Phenotype" means an observable or measurable expression of a gene or genes.

"Phenylketonuria" means an inborn error of amino acid metabolism, resulting in an inability to convert phenylalanine to tyrosine.

"Positive" means a laboratory result on an acceptable specimen which is designated as having high risk for disease to justify follow-up action.

"Repeat specimen" means a specimen collected because the previous specimen was unacceptable or previous test results were too early, or otherwise unreliable. The repeat specimen is a redrawn specimen.

"Repeat test" means a test ordered to be performed on a repeat specimen.

"Satisfactorily tested" means that the infant had newborn screening tests on an acceptable [specimen(s)] specimen or specimens with either negative or positive results, and the infant with positive results received appropriate follow-up with repeat or confirmatory tests to establish or disprove the presence of the disease.

"Unacceptable specimen" means any specimen which has been improperly collected, handled, or transported to the laboratory such that the specimen cannot be tested to yield acceptable results for the [test(s)] test or tests for which it was submitted.

"Urea cycle disorders" means a group of hereditary disorders, caused by enzymatic defects which result in a toxic accumulation of ammonia in the blood.

"Working day" means an official day of work for administrative programs of the department. [Eff 1/24/87; am and comp] (Auth: HRS §321-291) (Imp: HRS §321-291)

§11-143-4 Diseases required to be screened. All infants born in the State shall be satisfactorily tested for phenylketonuria, congenital hypothyroidism, congenital adrenal hyperplasia, galactosemia, biotinidase deficiency, hemoglobinopathies, [and] maple syrup urine disease, amino acid disorders, fatty acid oxidation disorders, organic acid disorders, and urea cycle disorders as described in this chapter. [Eff 1/24/87; am and comp] (Auth: HRS §321-291) (Imp: HRS §321-291)

§11-143-4.5 Fees and special fund. (a) Effective [July 1,1997] September 1,2003, the department shall collect a fee of [\$27] \$47 for each newborn screening kit. [The fee may not increase more than ten per cent

each year.] The department shall deposit the revenues into the newborn metabolic screening special fund.

(b) Kits requested for testing shall be prepaid by the hospital, laboratory, or birth attendant in the amount specified by the department.

(c) No infant born in Hawaii shall be denied newborn screening testing because of inability of the infant's parent or legal guardian to pay the fee for newborn screening testing. [Eff 6/5/97; am and comp
] (Auth: HRS §321-291) (Imp: HRS §321-291)

§11-143-5 Hospital, birth attendant, and physician responsibilities. (a) The newborn's attending physician or birth attendant and the hospital administrator shall be jointly responsible for assuring that each infant born or transferred to their care is satisfactorily tested as specified in section 11-143-4.

(b) Each hospital shall have written [policy] policies and procedures concerning the required testing of newborns for designated metabolic and other diseases.

(c) All newborns shall have a newborn screening specimen drawn before discharge from the hospital, or by seven days of age, [which ever] whichever comes first, and sent to the designated laboratory, except as provided for in [section 11-143-5(e).] subsection (e).

(d) The newborn shall not be discharged until the medical record is checked to assure that the newborn screening specimen has been collected.

(e) For newborns transferred from one hospital to another, the originating hospital shall assure that the newborn screening specimen is drawn. If the newborn is too premature or too sick to have a specimen drawn prior to transfer and a specimen is not obtained, the originating hospital shall be responsible for clearly documenting this, notifying the hospital to which the newborn is being transferred that a specimen has not been obtained, and reporting to the department as described in subsection (g).

(f) The hospital shall keep record summaries of infants born or transferred by month of birth, as to whether the newborn screening tests were done, the test results, and actions taken based on test results or missing results. These summaries shall be compiled monthly and sent to the department not later than thirty days after the end of the month.

(g) Not later than forty-eight hours after transfer, discharge, or death, the hospital shall report to the department, on a form provided by the department, the name of the newborn and parents or legal guardians' names, address, phone number, and physician's name for those newborns not tested prior to transfer, discharge, or death from the hospital.

(h) Each hospital shall have available for distribution to parents and legal guardians copies of the parent information brochure provided by the designated laboratory.

(i) Each hospital, physician, and birth attendant shall ensure compliance with section 11-143-6.

(j) Each physician shall follow the American Academy of Pediatrics newborn screening recommendations for repeat screening for infants whose initial specimens are obtained before twenty-four hours of age.

(k) The physician caring for the newborn who has a positive newborn screening test shall order confirmatory tests. The physician may request assistance from the department if the physician has difficulty contacting the family regarding the positive newborn screening test result.

(l) Hospital charges for the newborn screening tests shall be justifiable [and shall be reported to the department annually]. [Eff 1/24/87; am and comp

] (Auth: HRS §321-291) (Imp: HRS §321-291)

§11-143-6 Specimen collection. (a) All personnel responsible for collecting newborn screening specimens shall have read and become familiar with the department's Newborn Screening Practitioner's Manual or have participated in a training program, or both.

(b) Personnel collecting newborn screening specimens shall record the procedure and the fact that the newborn screening specimen has been collected in the infant's medical record.

(c) Specimen collection forms purchased from the department shall be used for all newborn screening specimens.

(d) All information requested on the specimen collection form shall be provided by personnel responsible for newborn screening specimen collection.

(e) For initial newborn screening [screening] tests, blood shall be taken from the newborn's heel or

dorsal hand vein and placed on the approved collection form.

(f) Specimen collection procedures shall follow the National Committee for Clinical Laboratory Standards, "Blood Collection on Filter Paper for Neonatal Screening Programs [-Second] - Second Edition".

(g) If a newborn is to be discharged prior to twenty-four hours of age, a newborn screening specimen shall be collected as close to discharge as possible, regardless of age and feeding history. If the initial newborn screening specimen is obtained before twenty-four hours of age, then a repeat specimen shall be obtained before fourteen days of age. Any new American Academy of Pediatrics newborn screening recommendations for specimens obtained before twenty-four hours of age shall supercede these recommendations.

(h) Newborns who require a blood transfusion or dialysis shall have a specimen collected prior to transfusion or dialysis. If a newborn screening specimen cannot be obtained before transfusion or dialysis, the physician shall ensure that a repeat specimen is obtained at the appropriate time when the specimen will reflect the infant's own metabolic processes and phenotype.

(i) All newborn screening specimens shall be sent to the designated laboratory within twenty-four hours of collection, except when mailing service is not available. When mailing service is not available on weekends and holidays, newborn screening specimens shall be sent to the designated laboratory on the first available mail pick-up day. [Eff 1/24/87; am and comp

] (Auth: HRS §321-291) (Imp: HRS §321-291)

§11-143-7 Parental notification and refusal. (a) Copies of the parent information brochure shall be available to parents, [legal] guardians, other persons having custody or control of the child, hospitals, physicians, birth attendants, birth registrars, nurses, and childbirth educators.

(b) The parent, [or legal] guardian, or other person having custody or control of the child shall be notified by the department of the need for repeat or confirmatory testing when the department is not able to obtain follow-up information from the physician.

(c) The parent, [or legal] guardian, or other person having custody or control of the child may refuse the newborn screening tests for [the parent or legal guardian's] their infant on the grounds that the newborn screening tests conflict with the [parent or legal guardian's] religious tenets and beliefs of the parent, guardian, or other person having custody or control of the child. The medical implications of that refusal shall be included on a special refusal form provided by the department. The refusal form shall be signed by the parent, [or the legal] guardian, or other person having custody or control of the child.

(d) A copy of the refusal form shall be retained in the newborn's medical record and a copy shall be sent to the department. [Eff 1/24/87; comp
] (Auth: HRS §321-291) (Imp: HRS §321-291)

§11-143-8 Home and non-institutional births. (a) For births occurring outside of a hospital, the birth attendant shall be responsible for assuring that an acceptable specimen is properly collected for testing as stipulated in section 11-43-6.

(b) For unattended, home, and non-institutional births, the department's birth registrar shall give the person registering the birth of the child a copy of the parent information brochure on newborn screening for metabolic and other diseases and send a copy of the birth certificate to the newborn screening program. [Eff 1/24/87; am and comp
] (Auth: HRS §321-291) (Imp: HRS §321-291)

§11-143-9 Laboratory responsibilities. (a) A laboratory wishing to offer the newborn screening tests shall submit a proposal for provision of newborn screening laboratory services, as specified by the department.

- (b) The designated laboratory shall:
- (1) Provide laboratory analysis for the initial newborn screening tests, for the repeat newborn screening tests, and confirmatory tests on positive screening results;
 - (2) Maintain a system of linking all test results with the appropriate newborn;
 - (3) Provide specimen collection forms and parent information brochures to hospitals,

- laboratories, birth attendants, and the department;
- (4) Provide practitioner's manuals to the department;
 - (5) Implement and follow procedures for keeping all specimens received under adequate storage conditions to allow for retesting for at least one year;
 - (6) Implement and follow record keeping procedures which include:
 - (A) Procedures for the logging of received specimens;
 - (B) Procedures for tracking repeat testing for previous unacceptable specimens and positive tests;
 - (C) Procedures for the reporting of all test results to the newborn's physician, hospital, and the department;
 - (D) Procedures for the reporting of positive tests and unacceptable specimens to the newborn's physician and the department as soon as possible but not later than seven working days after receipt of the specimen;
 - (7) Implement and follow a schedule of reporting to the department:
 - (A) Daily reports [to] that shall include as a minimum:
 - (i) Positive test results case report, including newborn's name, parents' name, address, phone number, and name of primary care provider;
 - (ii) Unacceptable specimen case report, including newborn's name, parents' name, address, phone number, and name of primary care provider;
 - (B) Monthly and annual reports [to] that shall include as a minimum:
 - (i) Collection status: Number of acceptable and unacceptable specimens by source;
 - (ii) Unduplicated number of infants tested by source;
 - (iii) Number of positive and negative tests by disorder;
 - [(iv) Number of confirmatory tests by disorder;

- (v) Number of confirmed cases by disorder;
- (vi)] (iv) Number of repeat tests for previous specimens collected for newborns less than twenty-four hours of age;
- (8) Maintain an internal quality assurance program;
- (9) Participate in an external proficiency testing and quality assurance program approved by the department;
- (10) Implement and follow procedures for specimen handling and laboratory testing during emergencies or other situations when the laboratory is [shutdown] shut down for more than three working days;
- (11) Provide an ongoing educational program to maintain and upgrade knowledge and skills of laboratory staff; and
- (12) Arrange for designated specialists in metabolic, hemoglobin, and endocrine disorders to be available for consultation regarding interpretation of tests results, and guidelines for care, treatment, and follow-up of infants with positive test results.

(c) Only the laboratory that has been selected in writing by the department as the designated laboratory shall be allowed to offer newborn screening testing.
[Eff 1/24/87; am and comp] (Auth: HRS §321-291) (Imp: HRS §321-291)

§11-143-10 Repealed [R 6/5/97]

§11-143-11 Penalty. The penalty for noncompliance with this chapter shall include an administrative fine as specified in section 321-20, H.R.S. [Eff 1/24/87; comp] (Auth: HRS §§321-20, 321-291) (Imp: HRS §§321-20, 321-291)

§11-143-12 Confidentiality. All information, including records, correspondence, and documents, specific to individual newborns, shall be confidential and shall be used solely for the purposes of medical

intervention, counseling, scientific research, or reporting. The infant's name shall be kept confidential. [Eff 1/24/87; comp]
(Auth: HRS §321-291) (Imp: HRS §321-291)

§11-143-13 Retention of [Records] records and [Related Data] related data. All information, including records, correspondence, documents, and related data, specific to individual newborns, shall be retained as specified in section 622-58, H.R.S. [Eff 6/5/97; am and comp] (Auth: HRS §321-291, HRS §622-58) (Imp: HRS §321-291, HRS §622-58)

§11-143-14 to 11-143-99 (Reserved).

§11-143-100 Severability. If any provision of this chapter, or the application thereof to any person or circumstance is held invalid, the invalidity does not affect other provisions or applications of this chapter that can be given effect without the invalid provision or application, and to this end the provisions of this chapter are severable." [Eff 1/24/87; comp] (Auth: HRS §321-291) (Imp: HRS §321-291)

2. Material, except source notes, to be repealed is bracketed. New material is underscored.

3. Additions to update source notes to reflect these amendments and compilation are not underscored.

4. These amendments to and compilation of chapter 11-143, Hawaii Administrative Rules, shall take effect ten days after filing with the Office of the Lieutenant Governor.

I certify that the foregoing are copies of the rules, drafted in the Ramseyer format pursuant to the requirements of section 91-4.1, Hawaii Revised Statutes, which were adopted on _____ and filed with the Office of the Lieutenant Governor.

CHIYOME L. FUKINO, M.D.
Director of Health

APPROVED AS TO FORM:

Deputy Attorney General